Complete Summary

GUIDELINE TITLE

Maternal phenylketonuria.

BIBLIOGRAPHIC SOURCE(S)

American Academy of Pediatrics, Section on Genetics. Maternal phenylketonuria. Pediatrics 2001 Feb;107(2):427-8. [19 references]

COMPLETE SUMMARY CONTENT

SCOPE

DISEASE/CONDITION(S)

Maternal phenylketonuria (PKU)

GUIDELINE CATEGORY

Counseling
Management
Prevention

CLINICAL SPECIALTY

Family Practice
Medical Genetics
Obstetrics and Gynecology
Pediatrics

INTENDED USERS

Advanced Practice Nurses
Nurses

Physician Assistants
Physicians

GUIDELINE OBJECTIVE(S)

To recommend counseling for girls and women of childbearing age with all forms of phenylketonuria regarding their risks for adverse fetal effects with uncontrolled blood phenylalanine levels during pregnancy

TARGET POPULATION

Women and girls of childbearing age with all forms of phenylketonuria, including hyperphenylalaninemia

INTERVENTIONS AND PRACTICES CONSIDERED

1. Counseling of girls and women of childbearing age with elevated phenylalanine levels concerning their risks for adverse fetal effects
2. Referral of women and girls with elevated phenylalanine levels to an experienced phenylketonuria treatment center for genetic and nutritional evaluation and counseling
3. Assistance in obtaining adequate means for birth control in women unable or unwilling to maintain optimal phenylalanine blood levels
4. Monitoring of phenylalanine blood levels before conception and during pregnancy
5. Ultrasonography to detect fetal abnormalities during pregnancy

MAJOR OUTCOMES CONSIDERED

- Blood phenylalanine levels of 120-360 micromoles per liter (2-6 milligrams per deciliter) or 60-250 micromoles per liter (1-4 milligrams per deciliter) during pregnancy
- Incidence of adverse fetal effects

METHODOLOGY

METHODS USED TO COLLECT/SELECT EVIDENCE

Searches of Electronic Databases

DESCRIPTION OF METHODS USED TO COLLECT/SELECT THE EVIDENCE

Not stated

NUMBER OF SOURCE DOCUMENTS

Not stated

METHODS USED TO ASSESS THE QUALITY AND STRENGTH OF THE EVIDENCE

Not stated

RATING SCHEME FOR THE STRENGTH OF THE EVIDENCE

Not applicable

METHODS USED TO ANALYZE THE EVIDENCE

Review

DESCRIPTION OF THE METHODS USED TO ANALYZE THE EVIDENCE

Not stated

METHODS USED TO FORMULATE THE RECOMMENDATIONS

Not stated

RATING SCHEME FOR THE STRENGTH OF THE RECOMMENDATIONS

Not applicable

COST ANALYSIS

A formal cost analysis was not performed and published cost analyses were not reviewed.

METHOD OF GUIDELINE VALIDATION

Peer Review

DESCRIPTION OF METHOD OF GUIDELINE VALIDATION

Not stated

RECOMMENDATIONS

MAJOR RECOMMENDATIONS

1. All girls and women of childbearing age with elevated phenylalanine levels, including those with phenylketonuria and milder forms of hyperphenylalaninemia, should be identified and counseled concerning their risks for maternal phenylketonuria fetal effects with uncontrolled blood phenylalanine levels during pregnancy. The pediatrician should include this information in anticipatory guidance counseling during adolescence for girls with phenylketonuria. The women and girls also should be referred to an experienced phenylketonuria treatment center for genetic and nutritional evaluation and counseling, optimally before contemplating pregnancy.

2. Women with hyperphenylalaninemia who are unable or unwilling to maintain blood phenylalanine levels in the range for optimum pregnancy outcome should be assisted to obtain adequate means for birth control, including tubal ligation if requested.
3. Women with hyperphenylalaninemia who conceive with blood phenylalanine levels greater than 250 to 360 micromoles per liter (4-6 milligrams per deciliter) should be counseled concerning the risks to the fetus and offered detailed ultrasonography to detect fetal abnormalities (e.g., growth retardation, congenital heart defects). Termination of pregnancy may be considered by those who conceive with blood phenylalanine levels that are known to be associated with a high fetal risk (>900 micromoles per liter [>14.9 milligrams per deciliter]). (Report of Medical Research Council Working Party on Phenylketonuria, 1993)
4. Women who give birth to children with features of maternal phenylketonuria fetal effects without a known cause should undergo blood testing for hyperphenylalaninemia.

CLINICAL ALGORITHM(S)

None provided

EVIDENCE SUPPORTING THE RECOMMENDATIONS

REFERENCES SUPPORTING THE RECOMMENDATIONS

References open in a new window

TYPE OF EVIDENCE SUPPORTING THE RECOMMENDATIONS

The type of evidence supporting each recommendation is not specifically stated.

BENEFITS/HARMS OF IMPLEMENTING THE GUIDELINE RECOMMENDATIONS

POTENTIAL BENEFITS

- Improved control of maternal phenylalanine levels during pregnancy
- Reduction of risk of adverse fetal effects

POTENTIAL HARMS

Not stated

QUALIFYING STATEMENTS

QUALIFYING STATEMENTS

The recommendations in this statement do not indicate an exclusive course of treatment or serve as a standard of medical care. Variations, taking into account individual circumstances, may be appropriate.

IMPLEMENTATION OF THE GUIDELINE

DESCRIPTION OF IMPLEMENTATION STRATEGY

An implementation strategy was not provided.

INSTITUTE OF MEDICINE (IOM) NATIONAL HEALTHCARE QUALITY REPORT CATEGORIES

IOM CARE NEED

Living with Illness
Staying Healthy

IOM DOMAIN

Effectiveness
Patient-centeredness

IDENTIFYING INFORMATION AND AVAILABILITY

BIBLIOGRAPHIC SOURCE(S)

American Academy of Pediatrics, Section on Genetics. Maternal phenylketonuria. Pediatrics 2001 Feb;107(2):427-8. [19 references]

ADAPTATION

Not applicable: The guideline was not adapted from another source.

DATE RELEASED

2001 Feb

GUIDELINE DEVELOPER(S)

American Academy of Pediatrics - Medical Specialty Society

SOURCE(S) OF FUNDING

American Academy of Pediatrics

GUIDELINE COMMITTEE

Committee on Genetics

COMPOSITION OF GROUP THAT AUTHORED THE GUIDELINE

Committee on Genetics, 2000-2001: Christopher Cunniff, MD, Chairperson; Jaime L. Frias, MD; Celia Kaye, MD, PhD; John B. Moeschler, MD; Susan R. Panny, MD; Tracy L. Trotter, MD

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Section Representative: H. Eugene Hoyme, MD (Section on Genetics)

Consultants: Rebecca S. Wappner, MD

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FINANCIAL DISCLOSURES/CONFLICTS OF INTEREST

Not stated

GUIDELINE STATUS

This is the current release of the guideline.

AAP Policies are reviewed every 3 years by the authoring body, at which time a recommendation is made that the policy be retired, revised, or reaffirmed without change. Until the Board of Directors approves a revision or reaffirmation, or retires a statement, the current policy remains in effect.

GUIDELINE AVAILABILITY

Electronic copies: Available from the American Academy of Pediatrics (AAP) Policy Web site.

Print copies: Available from AAP, 141 Northwest Point Blvd., P.O. Box 927, Elk Grove Village, IL 60009-0927.

AVAILABILITY OF COMPANION DOCUMENTS

None available

PATIENT RESOURCES

None available

NGC STATUS

This summary was completed by ECRI on October 17, 2001. The information was verified by the guideline developer as of December 5, 2001.

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